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A Rare Disease in Two Brothers.

By CHARLES HUNTER, Major C.A.M.C., M.D.

R. C. AND G. C., brothers, aged 10 and 8 years, of British parentage, were admitted into Winnipeg General Hospital on May 12, 1915. Father living, aged 48 years, strong and healthy, and of normal

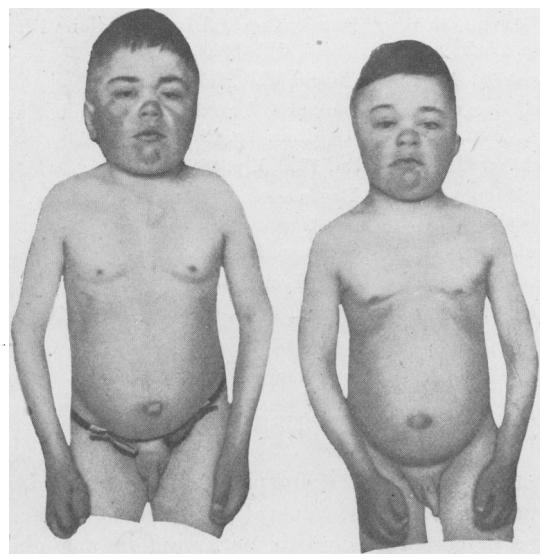

FIG. 1.

appearance. Mother died at the age of 45 years of kidney disease, while five months pregnant; had been previously a normal, healthy woman.

Their father's parents were cousins and had twelve children, one of these, when aged about 24 years, had an accident to his back and later went "insane with delusions about himself"; the others are healthy and their children are also healthy. The father knows his wife's family well; her parents, brothers and sisters were normal. She had, however, a deaf and dumb uncle.

The boys are the only living children. There was first a miscarriage at the age of 4 months; then full-term twins who died at birth—the confinement being difficult, and instrumental (these twins had, according to the father, large heads); then a miscarriage; and, lastly, death of the mother when pregnant five months.

The two children were full-term and were delivered without instruments; were both breast-fed and had no digestive disturbance in

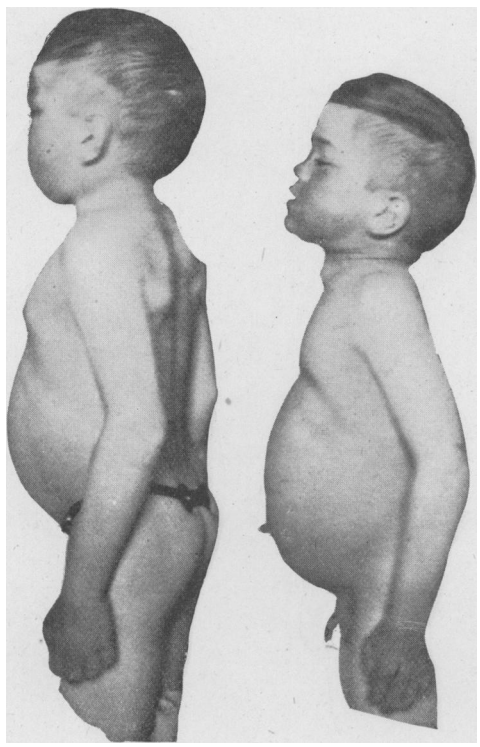

FIG. 2.

infancy; were both walking about the age of 17 months. The elder began to talk when aged about 1 year, is bright and intelligent, went to school at the age of 7 years and is in Grade 3. The younger was late in learning to talk and is still somewhat backward; he went to school a year ago and is making slow progress. They are good natured children on the whole, though the younger is at times a little "cranky"; they enjoy playing the ordinary games of childhood. They have been healthy in every way, apart from throat trouble. Both were operated

on for tonsils and adenoids; both are dull of hearing, and the father thinks this is getting slightly worse. They always get puffed when they run about. Both have had good appetites and regular bowels. Both had inguinal hernia; the younger was able to dispense with his truss three years ago, the elder still requires to wear one. They never had rheumatism nor growing pains.

Present condition: The children present an extraordinary appearance, and apart from their difference in size and one or two minor points to be

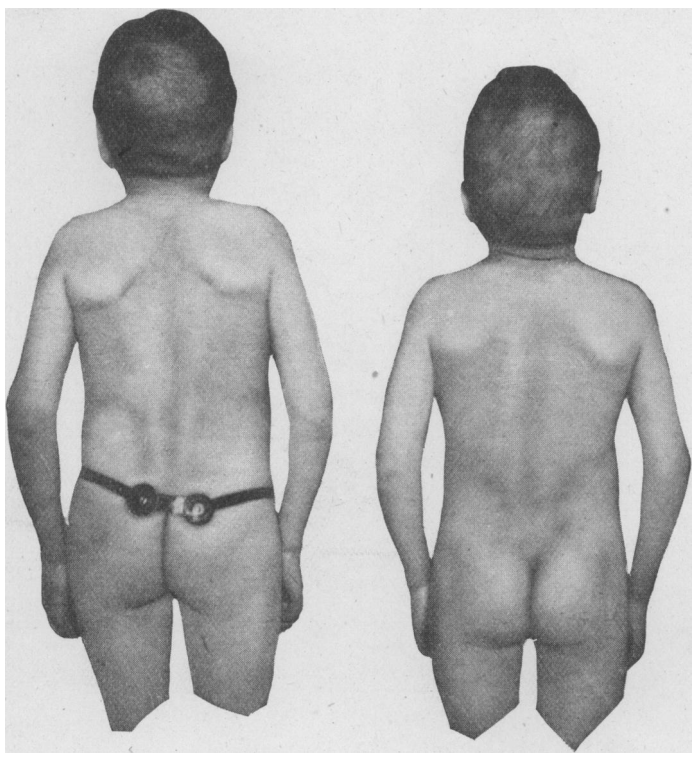

FIG. 3.

noted, they are as alike as two peas, so a common description may be given, the accompanying photographs (figs. 1 to 4), exhibiting the main features of interest. The children are undersized, 3 ft. 11 in. and 3ft. 9 in. (average, 4 ft. 4 in. and 4 ft.); weight, 56 lb. and 50 lb. (average, 66½ lb. and 54½ lb.); heads extremely large, measuring in greatest circumference 23 in. and 22 in. (average, 21 in. and 20½ in.). The head is curiously shaped, with very marked bulging of the squamous portion of the

temporal bone and of the frontal bones; the hair of the head rather thin and very harsh, especially in the younger. The face is very large, of deep burnt-red colour, as after much exposure, with a tinge of cyanosis in cheeks and lips; eyes very puffy; saddle nose, with large thick nostrils; thick lips, slightly open mouth, very large tongue; teeth good, but with irregular furrows and slightly spaced; very short neck, with slight enlargement of right lobe of thyroid in both. The chest is broad; abdomen very large and deep, greatly protruding, with small umbilical hernia; penis rather large. From behind, the scapulæ are seen to be placed extremely high, closely resembling a double Sprengel's deformity;

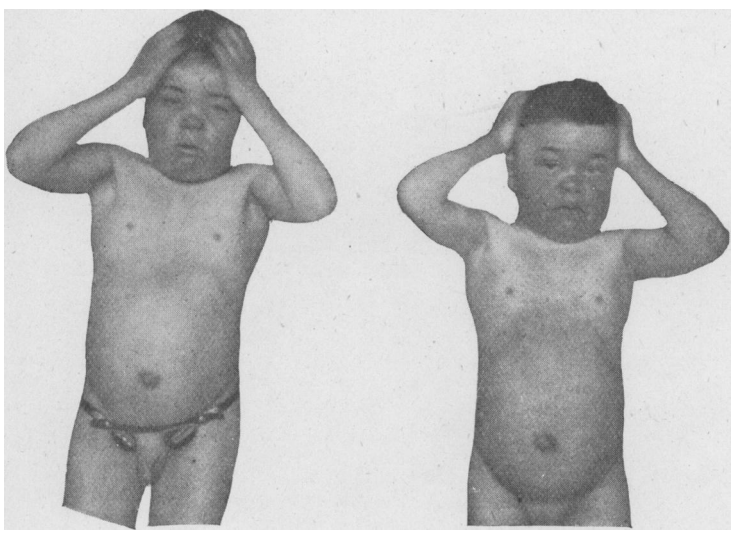

FIG. 4.

the neck is very short. The spinal column is straight, the natural curves being obliterated; the thighs are slightly bent, and the whole trunk inclines slightly forward. The arms are held somewhat abducted from the body and bent at the elbows so that the hands rest on the front of the thighs instead of on the side. The upper arm is disproportionately short in relation to the forearm, which is abnormally flat. The wrists are very thick; the hands very broad, short and thick; the fingers very short and bent. The knees are slightly flexed; both knees and ankles are thick, and the feet are broad, short and thick. The gait is very clumsy and stiff; the trunk is slightly bent forward and is held rigid. The normal extent of movement is curtailed in all

the joints of the extremities. The hands, particularly of the elder brother, have entirely lost their supple freedom of movement; they cannot be clenched; complete extension of the fingers is similarly defective, and even the movements possible are clumsy and stiff. The elbow and shoulder share in the general limitation of movement, and this is well shown in fig. 4, where the children are shown trying unsuccessfully to raise their hands above their heads. The photograph shows too the slightly greater freedom of movement possessed by the

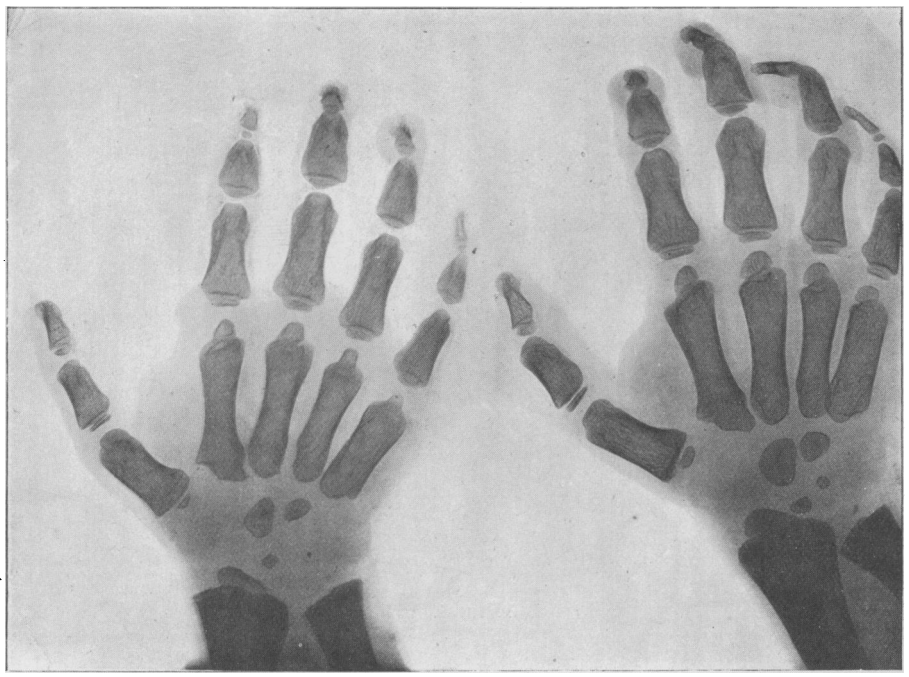

FIG. 5.

younger child. Active and passive movements of the spine are very much limited. The hip- and knee-joints have lost a little of their range of movement. The toes, while not deformed like the fingers, have completely lost the suppleness characteristic of youth. The skin of the trunk is smooth and not specially dry. The backs of the hands and fingers are deeply bronzed and the skin is there very thick and rough. In the younger child, over strips of skin $1\frac{1}{2}$ in. wide, extending from the angles of both scapulæ parallel to the ribs forward to the mid-axillary

lines, there are pinhead elevations, grouped closely and regularly, smooth of surface, normal in colour, and not unlike, though more superficial than, the lesions of cheiropompholyx. Some sixteen similar thickenings occur over an area of the size of 50 cents, in the upper part of the right arm. No pubic nor axillary hair. Nails normal, with crescents. The breathing is audible even at rest and becomes loud and puffing on exertion; the children are easily winded; in sleep, their

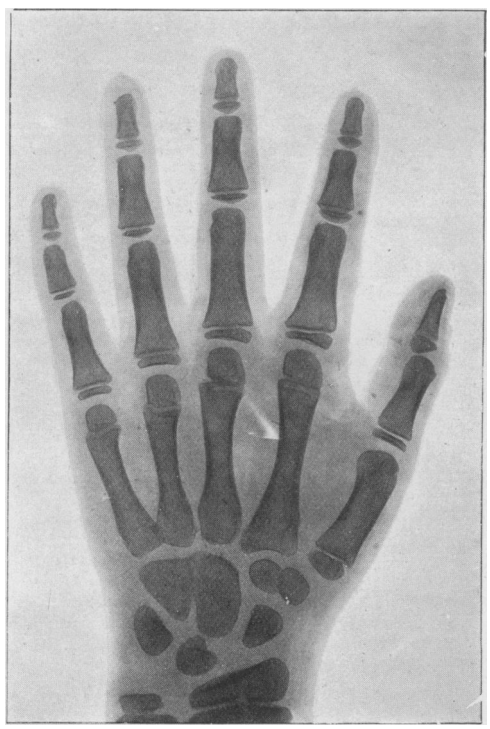

FIG. 6.

mouths remain wide open, the breathing being very laboured, uneasy and stertorous; there is an overhanging epiglottis; the lungs are normal. The heart in the younger is normal; in the elder, it is enlarged to the left, the apex beat being in fifth interspace just outside the nipple; there is a distinct diastolic murmur audible in the third and fourth left interspaces close to the sternum, the second sound at pulmonic and aortic areas being, however, clear; at the apex, a systolic murmur is conducted slightly towards the axilla. The elder seems, however, capable

of quite as much exertion as the younger, and like the younger he has only a tinge of cyanosis on cheeks and lips. Blood count in the elder: Hæmoglobin, 80 per cent.; red blood cells, 6,000,000; white cells, 7,000. The liver is very much enlarged in both, crossing in the case of the elder from the level of the right anterior superior spine to an inch above

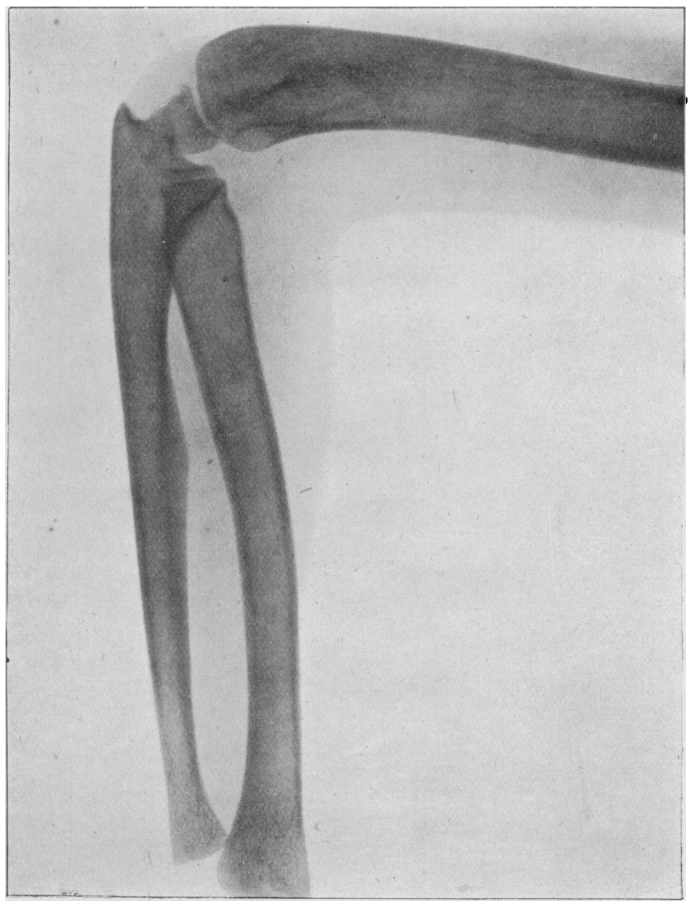

FIG 7.

the umbilicus, and in the younger, reaching a shade lower; the organ is smooth, not hard, the edge moderately thin, free from tenderness. The spleen is very much enlarged, reaching, in the elder fully two, and in the younger fully three, finger-breadths below the costal margin. The urine is normal. The testes are normal in size.

The children are bright and intelligent, particularly the elder, though both are hampered by distinct dullness of hearing. Speech is rather indistinct and they talked little, but they were under observation only two days, and during that time they were subjected to much examination. The father reported that they talked freely at home. General examination of the nervous system proved negative.

Wassermann reaction negative in both children and also in the father.

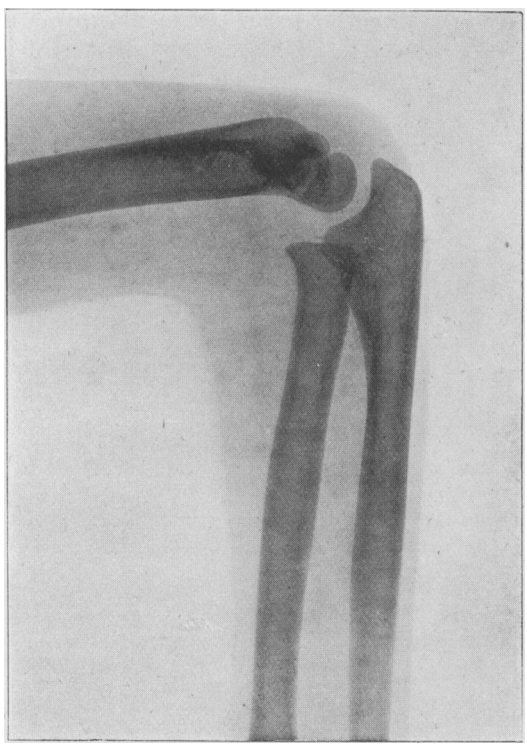

FIG. 8.

Dr. Prowse reported that both had some adenoids and that the whole of Waldeyer's ring was rather prominent; tympanic membranes retracted and somewhat hyperæmic. There were no signs of syphilitic disease about the nose and throat.

When the X-ray photographs are examined, we are struck with the abnormal thickness of all the bones and the pronounced irregular epiphyseal ossification. Fig. 5 shows the hand of the elder boy on the

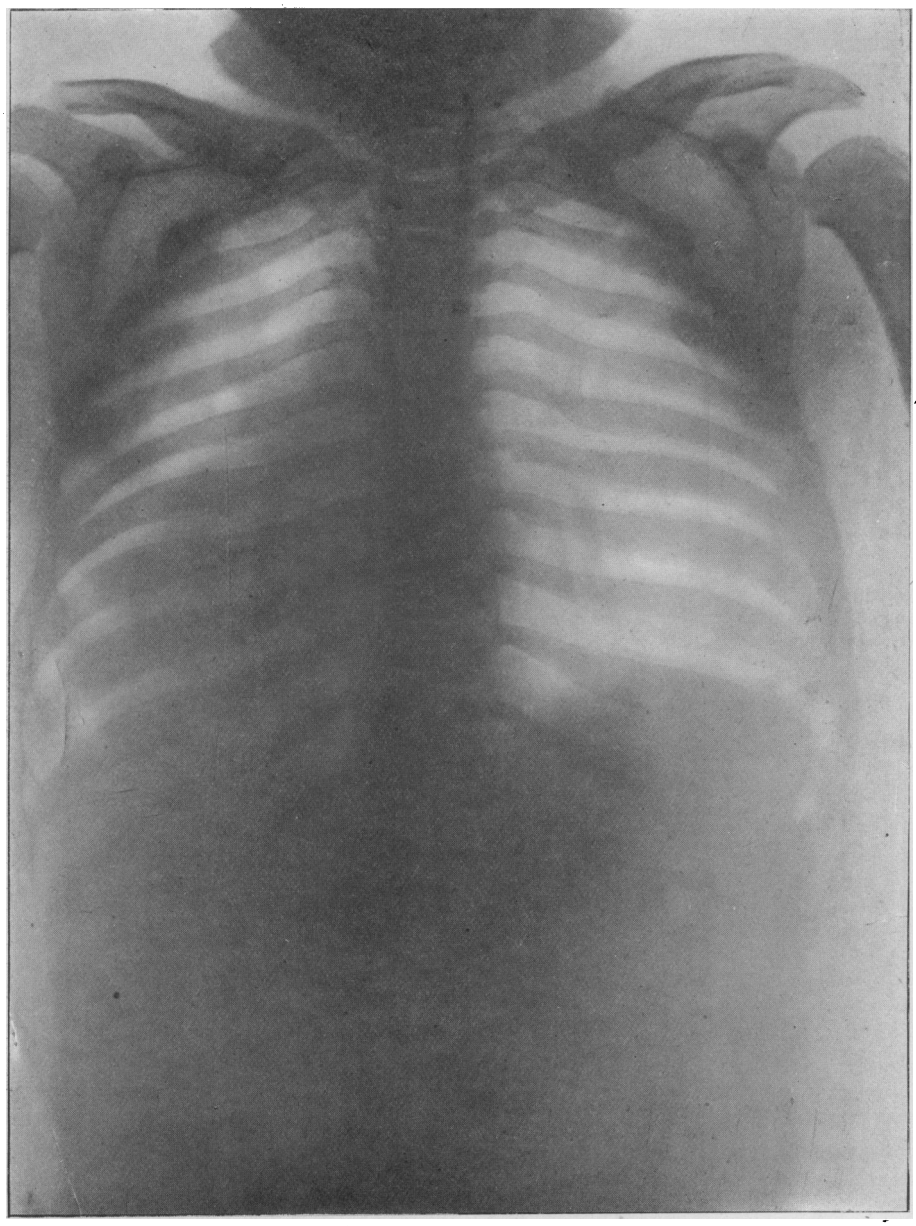

FIG. 9.

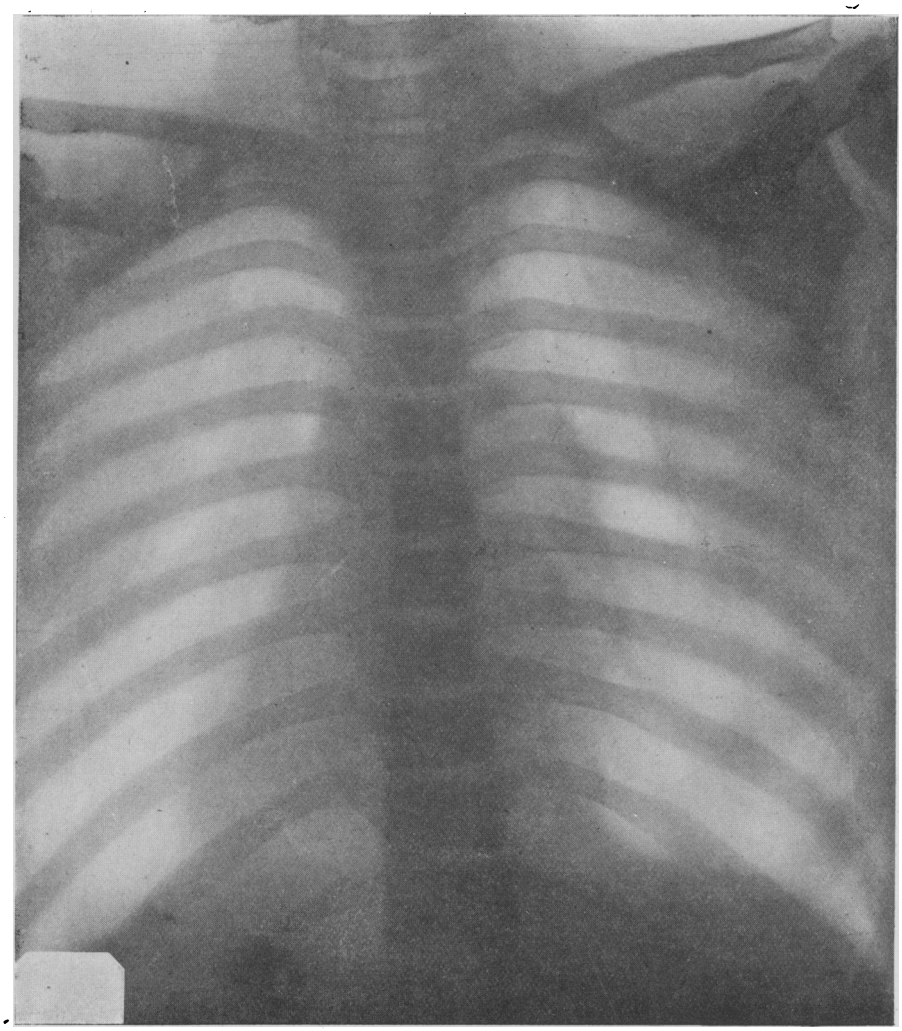

FIG. 10.

left and of the younger on the right as contrasted with the normal nine year old hand in fig. 6: the ossification of the carpus is distinctly delayed and the phalangeal and metacarpal bones are broad and short, with their epiphyseal ossification irregular and retarded. The difficulty experienced in straightening the fingers, especially in the case of the elder boy, is well shown. So in fig. 7, the general bulk of the bones

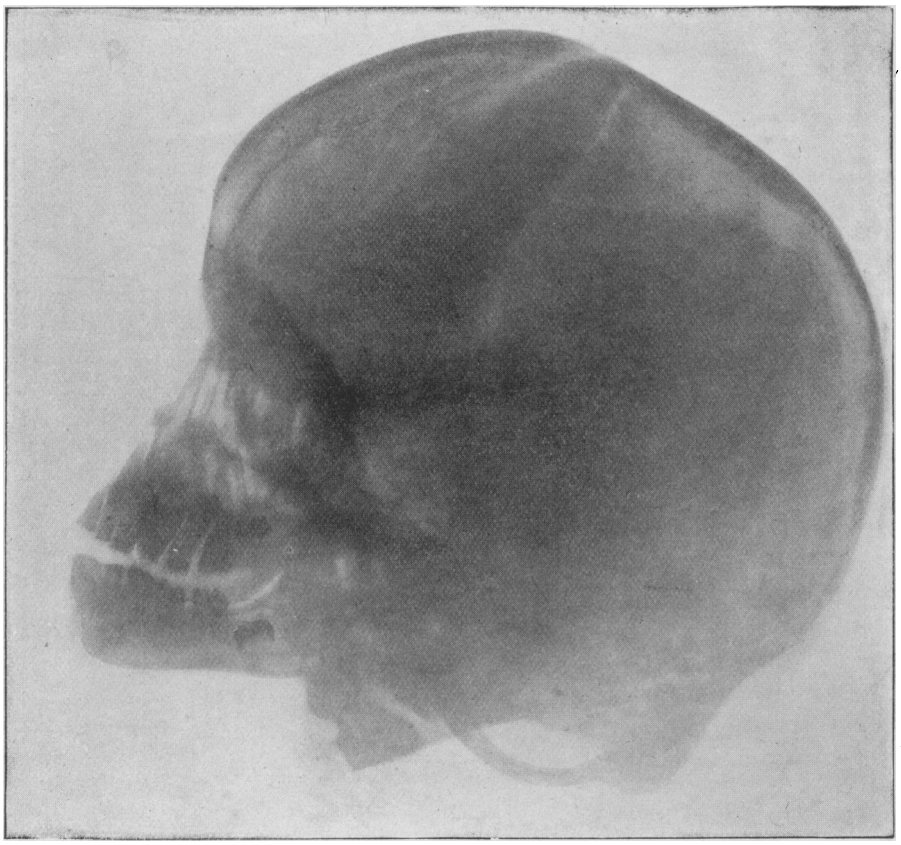

FIG. 11.

around the elbow is increased, with irregular epiphyseal ossification, as contrasted with the normal appearance as shown in fig. 8.

Fig. 9 again shows irregular epiphyseal ossification affecting both the head of the humerus and the glenoid cavity; the clavicles are enormously increased in bulk; the ribs are thickened in their transverse

diameter and are of a peculiar undulatory outline: the lateral surface of the vertebral bodies shows an irregular notched appearance and the high position of the scapulæ is well shown, as contrasted with the normal appearance as seen in fig. 10.

Fig. 11 shows the skull of the younger, and fig. 12 that of the elder boy, and in them we see the peculiar outline of the skull, its adult size, and its thickness of bone, with a sella turcica equal to the adult sella in size.

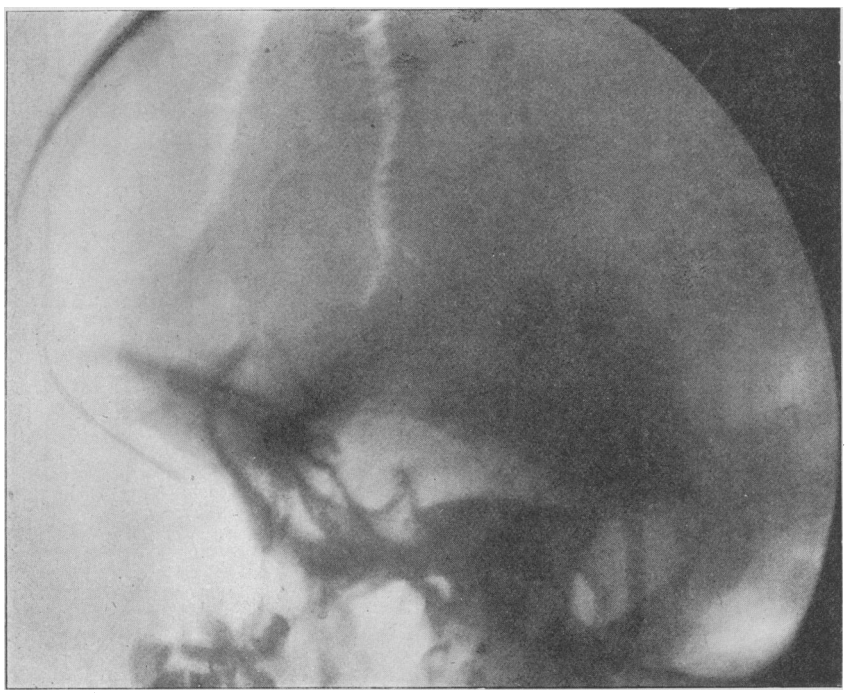

FIG. 12.

Owing to the limited library facilities of Winnipeg, I have been unable to find any reference to such a symptom complex.

DISCUSSION.

Dr. F. PARKES WEBER: I think everything in these boys points to the condition being an endocrine disturbance, but it would be at present wasting time to try to be more precise, as there are not yet sufficient data for the purpose. A remarkable feature in these two brothers is the "precociously

plethoric" appearance of their faces, such as is seen in some cases of hypernephroma in children. The appearance of the face and hands is well described by the author as that of a middle-aged farmer who is fond of malt liquor, and whose work naturally exposes him much to the weather.

Mr. BLUNDELL BANKART: I think that these are cases of multiple congenital defects of development. When viewed behind, these children present the appearance of bilateral Sprengel's deformity (congenital elevation of the scapula), a condition which is frequently associated with contracture of the shoulder-joint and defects of the vertebræ, ribs and other parts, as seen in these cases. The contracture of the fingers is another congenital condition which is often seen either alone or in conjunction with other contractures or defects. Sprengel's shoulder is attributed to an arrest of the normal descent of the scapula during intra-uterine development. It is difficult to see how any endocrine disturbance can be held responsible for such defects.